



KRT16 gene

keratin 16

Normal Function

The *KRT16* gene provides instructions for making a protein called keratin 16 or K16. Keratins are a group of tough, fibrous proteins that form the structural framework of certain cells, particularly cells that make up the skin, hair, and nails. Keratin 16 is produced in the nails, the skin on the palms of the hands and soles of the feet, and the oral mucosa that lines the inside of the mouth.

Keratin 16 partners with a similar protein, keratin 6a, to form molecules called keratin intermediate filaments. These filaments assemble into dense networks that provide strength and resilience to the skin, nails, and other tissues. Networks of keratin intermediate filaments protect these tissues from being damaged by friction and other everyday physical stresses. Keratin 16 is also among several keratins involved in wound healing.

Health Conditions Related to Genetic Changes

pachyonychia congenita

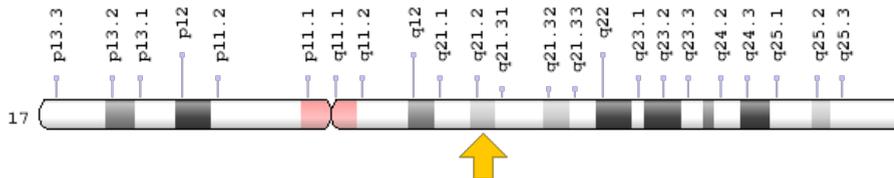
At least 19 mutations in the *KRT16* gene have been identified in people with pachyonychia congenita, a rare condition that primarily affects the nails and skin. In most cases, this condition becomes apparent within the first few months of life. Most of the *KRT16* gene mutations associated with pachyonychia congenita change single protein building blocks (amino acids) in keratin 16. A few mutations delete a small number of amino acids from the protein.

The *KRT16* gene mutations responsible for pachyonychia congenita change the structure of keratin 16, preventing it from interacting effectively with keratin 6a and interfering with the assembly of the keratin intermediate filament network. Without this network, skin cells become fragile and are easily damaged, making the skin less resistant to friction and minor trauma. Even normal activities such as walking can cause skin cells to break down, resulting in the formation of severe, painful blisters and calluses. Additionally, fragile skin cells may abnormally produce more keratin in response to damage, which makes the skin problems worse. Defective keratin 16 also disrupts the growth and function of other tissues, such as the hair follicles and nails, which explains why the signs and symptoms of pachyonychia congenita can also affect these other parts of the body.

Chromosomal Location

Cytogenetic Location: 17q21.2, which is the long (q) arm of chromosome 17 at position 21.2

Molecular Location: base pairs 41,609,778 to 41,612,827 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CK16
- cytokeratin 16
- cytokeratin-16
- K1C16_HUMAN
- K1CP
- K16
- keratin-16
- keratin 16 (focal non-epidermolytic palmoplantar keratoderma)
- keratin 16, type I
- keratin, type I cytoskeletal 16
- KRT16A
- NEPPK

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Keratin Filaments in Epithelial Cells (Image)
<https://www.ncbi.nlm.nih.gov/books/NBK26862/figure/A2988/>
- The Cell: A Molecular Approach (second edition, 2000): Intermediate Filaments
<https://www.ncbi.nlm.nih.gov/books/NBK9834/>

GeneReviews

- Pachyonychia Congenita
<https://www.ncbi.nlm.nih.gov/books/NBK1280>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28KRT16%5BTIAB%5D%29+OR+%28keratin+16%5BTIAB%5D%29+OR+%28K16%5Btiab%5D++AND+keratin%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- KERATIN 16, TYPE I
<http://omim.org/entry/148067>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_KRT16.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=KRT16%5Bgene%5D>
- HGNC Gene Family: Keratins, type I
<http://www.genenames.org/cgi-bin/genefamilies/set/608>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6423
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3868>
- UniProt
<http://www.uniprot.org/uniprot/P08779>

Sources for This Summary

- Fu T, Leachman SA, Wilson NJ, Smith FJ, Schwartz ME, Tang JY. Genotype-phenotype correlations among pachyonychia congenita patients with K16 mutations. *J Invest Dermatol*. 2011 May;131(5):1025-8. doi: 10.1038/jid.2010.373. Epub 2010 Dec 16.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21160496>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3775566/>
- McLean WH, Hansen CD, Eliason MJ, Smith FJ. The phenotypic and molecular genetic features of pachyonychia congenita. *J Invest Dermatol*. 2011 May;131(5):1015-7. doi: 10.1038/jid.2011.59. Epub 2011 Mar 24. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21430705>
- McLean WH, Rugg EL, Lunny DP, Morley SM, Lane EB, Swensson O, Dopping-Hepenstal PJ, Griffiths WA, Eady RA, Higgins C, et al. Keratin 16 and keratin 17 mutations cause pachyonychia congenita. *Nat Genet*. 1995 Mar;9(3):273-8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/7539673>
- Paladini RD, Takahashi K, Gant TM, Coulombe PA. cDNA cloning and bacterial expression of the human type I keratin 16. *Biochem Biophys Res Commun*. 1995 Oct 13;215(2):517-23.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/7487986>
- Smith FJ, Fisher MP, Healy E, Rees JL, Bonifas JM, Epstein EH Jr, Tan EM, Uitto J, McLean WH. Novel keratin 16 mutations and protein expression studies in pachyonychia congenita type 1 and focal palmoplantar keratoderma. *Exp Dermatol*. 2000 Jun;9(3):170-7.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/10839714>
- Smith FJ, Liao H, Cassidy AJ, Stewart A, Hamill KJ, Wood P, Joval I, van Steensel MA, Björck E, Callif-Daley F, Pals G, Collins P, Leachman SA, Munro CS, McLean WH. The genetic basis of pachyonychia congenita. *J Invest Dermatol Symp Proc*. 2005 Oct;10(1):21-30. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16250206>
- Terrinoni A, Smith FJ, Didona B, Canzona F, Paradisi M, Huber M, Hohl D, David A, Verloes A, Leigh IM, Munro CS, Melino G, McLean WH. Novel and recurrent mutations in the genes encoding keratins K6a, K16 and K17 in 13 cases of pachyonychia congenita. *J Invest Dermatol*. 2001 Dec; 117(6):1391-6.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11886499>
- Wilson NJ, O'Toole EA, Milstone LM, Hansen CD, Shepherd AA, Al-Asadi E, Schwartz ME, McLean WH, Sprecher E, Smith FJ. The molecular genetic analysis of the expanding pachyonychia congenita case collection. *Br J Dermatol*. 2014 Aug;171(2):343-55. doi: 10.1111/bjd.12958. Epub 2014 Aug 6.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24611874>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4282083/>

Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/KRT16>

Reviewed: December 2015
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services